



## GP1BB gene

glycoprotein Ib platelet beta subunit

### Normal Function

The *GP1BB* gene provides instructions for making a protein called glycoprotein 1b-beta (GPIb $\beta$ ). This protein is one piece (subunit) of a protein complex called GPIb-IX-V, which plays a role in blood clotting. GPIb-IX-V is found on the surface of small cell fragments called platelets, which circulate in blood and are an essential component of blood clots. The complex can attach (bind) to a protein called von Willebrand factor, fitting together like a lock and its key. Von Willebrand factor is found on the inside surface of blood vessels, particularly when there is an injury. Binding of the GPIb-IX-V complex to von Willebrand factor allows platelets to stick to the blood vessel wall at the site of the injury. These platelets form clots, plugging holes in the blood vessels to help stop bleeding.

To form the GPIb-IX-V complex, GPIb $\beta$  interacts with other protein subunits called GPIb-alpha, GPIX, and GPV, each of which is produced from a different gene. GPIb $\beta$  is essential for assembly of the complex at the platelet surface and helps stabilize the complex once it is formed.

### Health Conditions Related to Genetic Changes

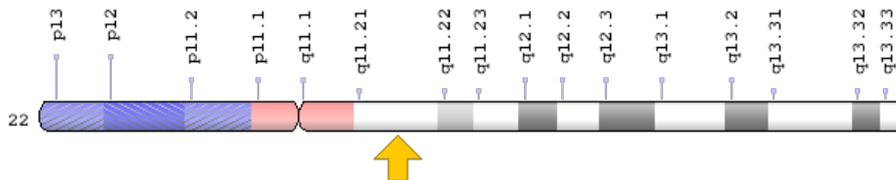
#### Bernard-Soulier syndrome

At least 32 *GP1BB* gene mutations have been found to cause Bernard-Soulier syndrome, a condition characterized by a reduced number of platelets that are larger than normal (macrothrombocytopenia) and excessive bleeding. These mutations lead to production of an altered GPIb $\beta$  subunit that is likely broken down too soon or that cannot get to the platelet surface. Lack of this subunit on the surface of platelets prevents formation of the GPIb-IX-V complex. Without GPIb-IX-V, platelets cannot come together at the site of an injury to form a clot, leading to the bleeding problems associated with Bernard-Soulier syndrome.

## Chromosomal Location

Cytogenetic Location: 22q11.21, which is the long (q) arm of chromosome 22 at position 11.21

Molecular Location: base pairs 19,723,543 to 19,724,774 on chromosome 22 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- antigen CD42b-beta
- BDPLT1
- BS
- CD42C
- glycoprotein Ib (platelet), beta polypeptide
- GP-Ib beta
- GPIBB
- GPIbbeta
- nuclear localization signal deleted in velocardiofacial syndrome
- platelet glycoprotein Ib beta chain precursor
- platelet membrane glycoprotein Ib beta
- truncated platelet membrane glycoprotein Ib beta

## Additional Information & Resources

### Educational Resources

- Clinical Methods: The History, Physical, and Laboratory Examinations (third edition, 1990): Excessive Bleeding and Bruising, Basic Science  
[https://www.ncbi.nlm.nih.gov/books/NBK253/#\\_A4391\\_](https://www.ncbi.nlm.nih.gov/books/NBK253/#_A4391_)
- Platelet-Vessel Wall Interactions in Hemostasis and Thrombosis (2010): Platelet Adhesion to Vascular Walls  
<https://www.ncbi.nlm.nih.gov/books/NBK53456/>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GP1BB%5BTIAB%5D%29+OR+%28glycoprotein+Ib+platelet+beta+subunit%5BTIAB%5D%29%29+OR+%28%28CD42C%5BTIAB%5D%29+OR+%28GP-Ib+beta%5BTIAB%5D%29+OR+%28GPIBB%5BTIAB%5D%29+OR+%28GPIbeta%5BTIAB%5D%29+OR+%28glycoprotein+Ib++,+beta+polypeptide%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

### OMIM

- GLYCOPROTEIN Ib, PLATELET, BETA POLYPEPTIDE  
<http://omim.org/entry/138720>

### Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=GP1BB%5Bgene%5D>
- HGNC Gene Family: CD molecules  
<http://www.genenames.org/cgi-bin/genefamilies/set/471>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=4440](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4440)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/2812>
- UniProt  
<http://www.uniprot.org/uniprot/P13224>

## Sources for This Summary

- OMIM: GLYCOPROTEIN Ib, PLATELET, BETA POLYPEPTIDE  
<http://omim.org/entry/138720>
- Geng H, Xu G, Ran Y, López JA, Peng Y. Platelet glycoprotein Ib beta/IX mediates glycoprotein Ib alpha localization to membrane lipid domain critical for von Willebrand factor interaction at high shear. *J Biol Chem*. 2011 Jun 17;286(24):21315-23. doi: 10.1074/jbc.M110.202549. Epub 2011 Apr 20.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/21507943>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3122191/>
- Li R, Emsley J. The organizing principle of the platelet glycoprotein Ib-IX-V complex. *J Thromb Haemost*. 2013 Apr;11(4):605-14. doi: 10.1111/jth.12144. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/23336709>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3696474/>
- McEwan PA, Yang W, Carr KH, Mo X, Zheng X, Li R, Emsley J. Quaternary organization of GPIb-IX complex and insights into Bernard-Soulier syndrome revealed by the structures of GPIb $\beta$  and a GPIb $\beta$ /GPIX chimera. *Blood*. 2011 Nov 10;118(19):5292-301. doi: 10.1182/blood-2011-05-356253. Epub 2011 Sep 8.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/21908432>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3217411/>
- Savoia A, Kunishima S, De Rocco D, Zieger B, Rand ML, Pujol-Moix N, Caliskan U, Tokgoz H, Pecci A, Noris P, Srivastava A, Ward C, Morel-Kopp MC, Alessi MC, Bellucci S, Beurrier P, de Maistre E, Favier R, Hézard N, Hurtaud-Roux MF, Latger-Cannard V, Lavenu-Bombled C, Proulle V, Meunier S, Négrier C, Nurden A, Randrianaivo H, Fabris F, Platokouki H, Rosenberg N, HadjKacem B, Heller PG, Karimi M, Balduini CL, Pastore A, Lanza F. Spectrum of the mutations in Bernard-Soulier syndrome. *Hum Mutat*. 2014 Sep;35(9):1033-45. doi: 10.1002/humu.22607. Epub 2014 Jul 15. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/24934643>
- Xu G, Shang D, Zhang Z, Shaw TS, Ran Y, López JA, Peng Y. The Transmembrane Domains of  $\beta$  and IX Subunits Mediate the Localization of the Platelet Glycoprotein Ib-IX Complex to the Glycosphingolipid-enriched Membrane Domain. *J Biol Chem*. 2015 Sep 4;290(36):22155-62. doi: 10.1074/jbc.M115.668145. Epub 2015 Jul 22.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/26203189>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4571966/>

---

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/GP1BB>

Reviewed: June 2016

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services